NEUROFIBROMATOSIS (VON RECKLINGHAUSEN'S DISEASE) OF THE VERTEBRAL COLUMN

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Although the occurrence of bone changes in von Recklinghausen's disease is uncommon, the incidence is difficult to determine, as mild cases are less likely to seek advice, particularly if the disease is already in the family. Holt and Wright (1948) thought it was greater than a previously estimated figure of 7%.

Of the bony deformities, scoliosis appears to be the commonest. Weiss (1921) described 15 cases of neurofibromatosis, all of whom had scoliosis. Brooks and Lehman (1924) studied seven cases with bone changes and found that all had scoliosis. They suggested that the lesion began with a neurofibroma of the periosteal nerve, and that this set up a reaction in the vertebra with bone destruction and regeneration. If actively bone-forming periosteum covers the tumour, a thin shell of bone forms over it and gives a cystic appearance. Weber (1930) described further cases and discussed various aspects of neurofibromatosis. Hagelstam (1946) reviewed the literature and collected 37 cases with deformity of the spine: two were in the cervical region, 20 in the upper and mid-dorsal regions and 15 in the dorsolumbar spines. Holt and Wright (1948) reviewed 127 cases of neurofibromatosis of which 29% showed skeletal involvement. The bone lesions consisted of erosive defects, scoliosis, growth disorders, bowing and pseudoarthrosis of the lower leg, intrasosseous cystic lesions and associated anomalies. McCarron (1950) found 19 cases of scoliosis in 43 cases with bone involvement.

Below are case reports of five children with neurofibromatosis of the spine.

Case Reports

Case 1. This girl was first seen at the age of 16 months on account of a 'bump in the back' which had been noticed from the age of 3 months. She was able to say several words with meaning and had learnt to sit without support at 10 months. She was unable to walk. The father, who was the youngest of 17 children, and his father had von Recklinghausen's disease, but as far as we could tell the father's siblings were unaffected. We did not, however, examine them. Our patient was one of two children, both of whom had the disease.

On examination the girl presented the typical appearance of von Recklinghausen's disease with involvement of the vertebrae (Figs. 1 and 2). There were large pigmented and nodular areas in the groins (Fig. 3) and a severe kyphosis of the upper lumbar spine with chest deformity. Radiographs showed upper lumbar scoliosis with increased density and translucent areas in the anterior and posterior surfaces of the vertebrae (Fig. 4). There was considerable widening of the neural canal, suggesting the presence of a tumour mass. There was a spastic paraplegia with exaggerated tendon jerks, bilateral ankle clonus and a bilateral extensor plantar response. The neurosurgeon was not prepared to operate.

The girl subsequently put words together into sentences at 2 years and walked without help at 2 years and 3 months. At the age of 5 years she developed a headache, mainly occipital, and ataxia. She became drowsy and could no longer walk. On examination bilateral papilloedema was found, and a diagnosis of intracranial neurofibroma was made. She died at home.

FIG. 1.—Case 1, aged 16 months. FIG. 2.—Case 1, aged 4 years.
Fig. 3.—Case 1, showing pigmented and nodular areas in groins.

Fig. 4.—Case 1.

Fig. 5.—Case 2.

Fig. 6.—Case 2, close-up photograph of affected skin.
Case 2. This girl was under the care of an orthopaedic surgeon from the age of 6 months on account of dorsolumbar kyphosis. She was referred to a paediatrician at the age of 12 years because of cyanosis and dyspnoea on exertion. She had been brought up in a children's home from the age of 6 months having been neglected by the parents. The family history was unknown.

On examination at the age of 12 years there was a severe kyphosis maximal in the lower thoracic region (Fig. 5). There was widespread brown discoloured skin of the skin involving the lower half of the trunk, buttocks and part of the thighs. The skin in these areas was raised and pedunculated (Fig. 6). No other abnormal physical signs were found.

A radiograph of the vertebrae showed exaggerated pedicles (Fig. 7) and that of the left femur showed patchy calcification and cortical destruction (Fig. 8). A chest radiograph was normal and it was concluded that the respiratory symptoms were due to thoracic deformity. Skin biopsy confirmed the diagnosis of neurofibromatosis.

Case 3. (Figs. 9-11.) Swellings of the neck were noticed in this boy at 11 months. At the age of 14 months he was seen at the Leeds General Infirmary. There was a bilateral cervical adenitis with mottling of the skin. A radiograph showed collapse of the body of the fourth cervical vertebra. The tuberculin test was negative. Biopsy of the gland mass gave the appearance of a neurofibroma.

He was immobilized on a plaster of paris bed with headpiece from 2 years 2 months to 5 years 10 months, by which time the neck was grossly swollen by a nodular mass which in places infiltrated and reddened the skin. The area was generally freckled and pigmented and on the trunk there were café-au-lait patches. Radio-

logically the condition of the neck was remarkable in that between the trachea and vertebral column there was a mass which had not pushed the trachea forward but had occupied a concavity of the cervical kyphosis as if it were responsible for the condition of the latter (Fig. 11). There had, however, been no specific change in the bones of the cervical vertebrae. An attempt was made to
excise the mass at the age of 6 years 1 month but this was unsuccessful. The boy subsequently attended a school for physically handicapped children.

At the age of 15 years his height was 48½ in. (1·21 m.) and his weight 48 lb. (21·8 kg.). He could walk fairly well but did not run. His appetite was good and he went to camp with the scouts.

FAMILY HISTORY. The mother felt on her face and body numerous painless nodules resembling neurofibromata. She remembered that they had appeared after an attack of 'erysipelas' at the age of 16.

Case 4. (Figs. 12-14.) Light brown spots were noticed on the trunk and thighs of this child between the ages of 1 and 3 years. They became more numerous and at the age of 6 years it was noticed that the back was curved.

On examination there were many café-au-lait macules up to the size of 3 cm., mainly on the trunk. There was a mid-dorsal scoliosis with convexity to the right. This could be passively but not actively reduced.

There was a gradual increase in the deformity in spite of orthopaedic treatment.
FAMILY HISTORY. The father had had brown spots, as had a paternal uncle and the paternal uncle's only daughter.

Case 5. A lump was noticed on the left side of the chest in the region of the nipple when the child was 16 months old (Fig. 15). When she was 2 years old a lump was noticed on the right side of the chest posteriorly (Fig. 16).

Examination at the age of 2 years showed that there was a scoliosis with the upper convexity to the left, the lower convexity to the right (Fig. 17). This largely disappeared when the child was lifted up. Over the trunk several café-au-lait patches were seen ranging from the size of a florin to a pea. No other skeletal deformity was noticed.

There was a gradual increase of the deformity in spite of physiotherapy.

FAMILY HISTORY. The mother was alleged to have been born with, or at all events to have developed shortly after birth, many coloured patches like those on the child. After the patient's birth soft lumps had appeared on her body, too.

Discussion

The changes in the vertebral column are of grave significance. Apart from a tendency to undergo malignant change, the lesions are progressive over at least several years, reducing the patient to a cripple. The diagnosis of neurofibromatosis may not be obvious if the skin lesions are minimal. In infancy there may be only one or two café-au-lait spots and several years may elapse before they become more numerous and prominent. While large subcutaneous masses are relatively frequent in infancy, the small polyp so characteristic in the adult is usually absent. The mother of Case 3 remembered that her nodules appeared at the age of 16 following an attack of erysipelas. The mother of Case 5 noticed the 'soft lumps' on her body after the birth of the patient.

In view of the grave prognosis, the diagnosis of neurofibromatosis should be considered in any case of unexplained scoliosis.

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